



DLL3 gene

delta like canonical Notch ligand 3

Normal Function

The *DLL3* gene provides instructions for making a protein that helps control (regulate) the Notch pathway, an important pathway in embryonic development. The Notch pathway plays a critical role in the development of vertebrae. Specifically, the *DLL3* protein and the Notch pathway are involved in separating future vertebrae from one another during early development, in a complex process called somite segmentation. Although the exact mechanism of somite segmentation is unclear, it appears to require the activity of several proteins in the Notch pathway, including the *NOTCH1* protein, to be turned on and off (oscillate) in a specific pattern.

The *DLL3* protein regulates the activity of the *NOTCH1* protein. The *DLL3* protein attaches (binds) to the inactive *NOTCH1* protein and isolates (sequesters) it or marks it to be broken down so that it cannot be activated.

Health Conditions Related to Genetic Changes

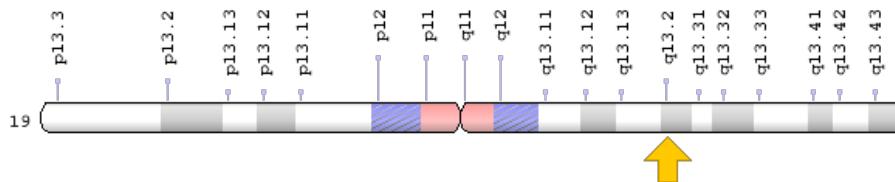
spondylocostal dysostosis

At least 20 mutations in the *DLL3* gene have been found to cause spondylocostal dysostosis type 1, the most common type of spondylocostal dysostosis. This condition is characterized by the abnormal development of bones in the spine and ribs. The known mutations in the *DLL3* gene prevent the production of any protein or lead to the production of an abnormally short, nonfunctional protein. When the *DLL3* protein is nonfunctional or absent, the *NOTCH1* protein is abnormally active and does not oscillate, so somite segmentation does not occur properly. This results in the malformation and fusion of the bones of the spine and ribs seen in spondylocostal dysostosis type 1.

Chromosomal Location

Cytogenetic Location: 19q13.2, which is the long (q) arm of chromosome 19 at position 13.2

Molecular Location: base pairs 39,498,917 to 39,508,481 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- delta-like 3 (Drosophila)
- delta-like protein 3
- delta-like protein 3 isoform 1 precursor
- delta-like protein 3 isoform 2 precursor
- delta3
- DLL3_HUMAN
- drosophila Delta homolog 3
- SCDO1

Additional Information & Resources

GeneReviews

- Spondylocostal Dysostosis, Autosomal Recessive
<https://www.ncbi.nlm.nih.gov/books/NBK8828>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DLL3%5BTIAB%5D%29+OR+%28delta-like+3%5BTIAB%5D%29%29+OR+%28SCDO1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- DELTA-LIKE 3
<http://omim.org/entry/602768>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_DLL3.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=DLL3%5Bgene%5D>
- GENATLAS
<http://genatlas.medecine.univ-paris5.fr/fiche.php?n=10830>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2909
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/10683>
- UniProt
<http://www.uniprot.org/uniprot/Q9NYJ7>

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